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Human Chromosome 16 Phospholipin-like Polypeptide

~~HUMAN CHROMOSOME 15 AND 16 BARDET-BIEDL SYNDROME  
POLYNUCLEOTIDES AND POLYPEPTIDES AND METHODS OF USE~~

TECHNICAL FIELD

This invention relates to the identification and recombinant expression of  
5 human chromosome 15 and 16 Bardet-Biedl Syndrome Region (BBSR) proteins.

BACKGROUND OF THE INVENTION

Bardet-Biedl Syndrome (BBS) is a clinically and genetically heterogeneous  
autosomal recessive disorder characterized by obesity, polydactyly, hypogenitalism, retinal  
degeneration, mental retardation and heart and kidney abnormalities.

10 Elbedour et al. (1994) *Am. J. Med. Genet.* 52(2):164-169 have reported  
hypertrophy of the interventricular septum and dilated cardiomyopathy, in addition to other  
previously reported congenital heart defects associated with BBS.

BBS has been mapped to loci on several human chromosomes including  
chromosomes 3, 11, 15 and 16. These loci include the 3p12, 11q13, 15q22 and the 16q21  
15 chromosomal sites; also referred to as the BBS3, BBS1, BBS4 and BBS2 loci, respectively.  
(Bruford et al. (1997) *Genomics* 41(1):93-9; Leppert et al. (1994) *Nature Genet.* 7:108-  
112; Carmi et al. (1995) *Hum. Mol. Genet.* 4:9-13; Kwitek-Black et al. (1993) *Nature*  
*Genet.* 5:392-396; Sheffield et al. (1994) *Hum. Mol. Genet.* 3:1331-1335; Beales et al.  
(1997) *J. Med. Genet.* 34(2):92-8).

20 Attempts to associate particular phenotypes with particular BBS loci have  
been reported. For example, Beales et al. (1997) *J. Med. Genet.* 34(2):92-8, reported that  
affected subjects linked to the BBS2 and 4 loci were significantly shorter than their parents,  
while those linked to the BBS1 locus were taller, indicating possible role for various BBS  
genes in influencing growth characteristics such as height. Carmi et al. (1995) *Hum. Mol.*  
25 *Genet.* 4:9-13, reported that BBS3 is associated with polydactyly of all four limbs while